



CLINICAL POLICY AND PROCEDURE MANUAL

Policy Number: PA.249.CC
Last Review Date: 11/16/2023
Effective Date: 12/01/2023

PA.249.CC Noninvasive Genetic Testing During Pregnancy

Summary:

ACOG estimates that 6-11% of stillbirths and neonatal deaths result from aneuploidies (fetus with missing or extra chromosomes). Most aneuploidies involve the presence of an extra chromosome, also referred to as trisomy.

Down syndrome, which is most commonly caused by trisomy 21 (T21), is routinely evaluated as the standard of care for the majority of the 4 million women who give birth each year in the United States. Conventional screening tests typically involve measurement of blood serum markers in conjunction with ultrasound followed by recommendation for diagnostic invasive procedures for abnormal results from screening.

First trimester combined screening (FTS) and integrated screening (INT) have the best screening performance, yet still only have T21 detection rates of 82-87% and 88-95%, respectively, at false positive rates of 5%. Invasive testing with amniocentesis or CVS is highly accurate but has up to a 3% risk of procedure related miscarriage. The reported complication rates have come down in the last 10 years, but there is still some procedure-related risk.

A prenatal test that evaluates cell-free DNA (cfDNA) in maternal blood has been shown to be highly accurate, with T21 detection rates >99%, 98% detection rate for fetal trisomy 18 and 99% detection rate for fetal trisomy 13 with a combined false positive rate of 0.13%.

cfDNA testing, can detect more T21 cases and at the same time reduce unnecessary invasive procedures and in turn fewer procedure related fetal losses. cfDNA testing, when used as a follow-up test for an abnormal result from the FTS or INT screening test can spare the vast majority of the 5% of women with false positive results from undergoing invasive diagnostic testing. Any woman with an abnormal result from



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cfDNA test should undergo diagnostic testing by amniocentesis or chorionic villus sampling.

Clinical Criteria:

CountyCare considers cell-free fetal DNA-based prenatal screening for fetal aneuploidy (trisomy 13, 18, and 21) medically necessary for women with a current single or twin gestation pregnancy.

OR

Cell-free fetal DNA-based prenatal testing for fetal sex determination is considered medically necessary for singleton pregnancies at increased risk of a sex (X)-linked condition or congenital adrenal hyperplasia.

Limitations:

Cell-free fetal DNA-based prenatal screening for fetal aneuploidy (trisomy 13, 18, 21) is considered not medically necessary for individuals not meeting the criteria above, including pregnancies involving 3 or more fetuses.

OR

Cell-free fetal DNA-based prenatal screening for fetal aneuploidy (trisomy 13, 18, and 21) in twin pregnancies is considered not medically necessary when the current pregnancy is affected by fetal demise, vanishing twin, or one or more anomaly detected in one or both of the twins.

OR

Cell-free fetal DNA-based prenatal testing for fetal sex determination is considered not medically necessary for pregnancies without an increased risk of a sex (X)-linked condition or congenital adrenal hyperplasia.

OR

Cell-free fetal DNA-based prenatal testing is considered not medically necessary for all other indications, including testing for microdeletion syndromes.

Cell-free fetal DNA-based screening for indications other than those listed in above clinical coverage criteria will not be covered.

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Only one cell-free DNA testing will be covered per pregnancy. Only one screening modality will be covered per pregnancy.

In patients with a positive screen, providers should consider evaluation with a maternal fetal medicine specialist, genetic counseling, comprehensive ultrasound and diagnostic testing.

Applicable Codes:

CPT codes covered if selection criteria are met:	
Code	Description
81479	Unlisted molecular pathology procedure
81507	Fetal aneuploidy (trisomy 21, 18, and 13) DNA sequence analysis of selected regions using maternal plasma, algorithm reported as a risk score for each trisomy

References:

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2. Hayes GTE Overview. Cell-Free DNA (cfDNA) [Formerly NIPS, NIPT] Screening for Fetal Rare Autosomal Trisomies. Annual Review Date: October 17, 2022.
3. Hayes GTE Overview. Cell-Free DNA (cfDNA) [Formerly NIPS, NIPT] Screening for Fetal Chromosomal Copy Number Variants. Annual Review Date: March 30, 2023.
4. Hayes GTE Overview. Cell-Free DNA (cfDNA) [Formerly NIPS, NIPT] Screening for Fetal Sex Chromosome Aneuploidy. Annual Review Date: October 20, 2022.
5. Hayes GTE Report. Cell-Free DNA (cfDNA) [Formerly NIPS, NIPT] Screening for Fetal Trisomy 21, 18, and 13 in Low-Risk Women with Singleton Pregnancy. Annual Review Date: April 25, 2023.
6. Hayes GTE Report. Cell-Free DNA (cfDNA) [Formerly NIPS, NIPT] Screening for Fetal Trisomy 21, 18, and 13 in High-Risk Women. Annual Review: February 11, 2022.
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10. National Institutes of Health, National Library of Medicine. Genetics Home Reference: Down Syndrome. Last Updated: June 1, 2020.
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11. National Institutes of Health, National Library of Medicine. Medline Plus: Quadruple screen test. Reviewed: 01/10/2022.
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13. National Institutes of Health, National Library of Medicine. PubMed Health. A.D.A.M. Medical Encyclopedia: Chorionic Villus Sampling. Reviewed; 11/10/2022.
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14. Scott F, Peters H, Boogert T, et al. The loss rates for invasive prenatal testing in a specialised obstetric ultrasound practice. Aust N Z J Obstet Gynaecol. 2002 Feb;42(1):55-58.
<http://www.ncbi.nlm.nih.gov/pubmed/11926642>

15. Screening for Fetal Chromosomal Abnormalities: ACOG Practice Bulletin, Number 226 American College of Obstetricians and Gynecologists’ Committee on Practice Bulletins—Obstetrics; Committee on Genetics; Society for Maternal-Fetal Medicine.
<https://www.acog.org/clinical/clinical-guidance/practice-bulletin/articles/2020/10/screening-for-fetal-chromosomal-abnormalities>

Revision History:

Revision	Date
Policy Created	April 2021
Approved	June 2021
Renumbered Policy as it was originally assigned a duplicate number, removed code 81420 as it no longer requires prior auth	October 2022
Updated dates in Reference #s 3 through	November 2022

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7; Replaced the links in Reference #s 14 and 16; minor formatting update to Reference #15	
Updated Evolent Logo, changed Evolent Health to Evolent; formatting updates throughout the policy; format update to Reference #1; updated Annual Review Date in Reference #s 3 and 5; updated Last Updated Date in References #s 8, 9, 10, 11, 12, 13; format updates to Reference #s 14 and 15	November, 2023

Disclaimer:

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